



CRTAP gene

cartilage associated protein

Normal Function

The *CRTAP* gene provides instructions for making a protein called cartilage associated protein. While the specific function of this protein is not known, it plays an important role in normal bone development. Cartilage associated protein works with two other proteins, leprecan and cyclophilin B, as part of a complex that helps process certain forms of collagen. Collagens are proteins that provide strength, support, and the ability to stretch (elasticity) to many body tissues.

The complex containing cartilage associated protein modifies a protein building block (amino acid) called proline in collagen molecules. This modification, which is known as proline 3-hydroxylation, appears to be critical for the normal folding and assembly of collagen. It also may be important for releasing collagen molecules into the spaces around cells (the extracellular matrix). The secretion of collagen from cells is necessary for the proper formation of connective tissues, such as bones, tendons, and cartilage, that form the body's supportive framework.

Health Conditions Related to Genetic Changes

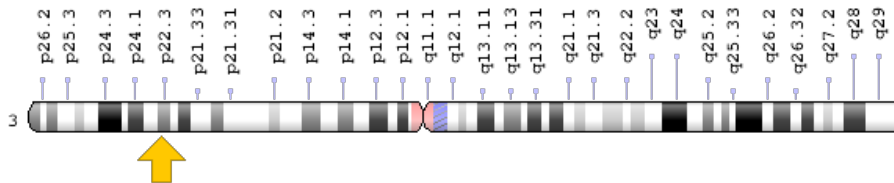
osteogenesis imperfecta

At least five mutations in the *CRTAP* gene are responsible for a rare type of osteogenesis imperfecta that is usually classified as type VII. Several of these mutations prevent cells from producing any cartilage associated protein. Without this protein, bones and other connective tissues do not form properly, leading to a very severe form of the disorder. Another mutation in the *CRTAP* gene greatly reduces the amount of cartilage associated protein produced, which disrupts the normal formation of collagen. This genetic change causes less severe signs and symptoms of osteogenesis imperfecta.

Chromosomal Location

Cytogenetic Location: 3p22.3, which is the short (p) arm of chromosome 3 at position 22.3

Molecular Location: base pairs 33,113,958 to 33,147,773 on chromosome 3 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- cartilage-associated protein
- CASP
- CRTAP_HUMAN

Additional Information & Resources

Educational Resources

- Howard Hughes Medical Institute: Genetic Mutation Explains Form of Brittle Bone Disease (October 20, 2006)
<http://www.hhmi.org/news/genetic-mutation-explains-form-brittle-bone-disease>
- Molecular Biology of the Cell (fourth edition, 2002): Collagens Are the Major Proteins of the Extracellular Matrix
<https://www.ncbi.nlm.nih.gov/books/NBK26810/#A3551>
- Molecular Cell Biology (fourth edition, 2000): Collagen: The Fibrous Proteins of the Matrix
<https://www.ncbi.nlm.nih.gov/books/NBK21582/>
- The Cell: A Molecular Approach (second edition, 2000): Collagen fibrils (figure)
<https://www.ncbi.nlm.nih.gov/books/NBK9874/?rendertype=figure&id=A2050>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28CRTAP%5BTIAB%5D%29+OR+%28cartilage+associated+protein%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

OMIM

- CARTILAGE-ASSOCIATED PROTEIN
<http://omim.org/entry/605497>

Research Resources

- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=CRTAP%5Bgene%5D>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=2379
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/10491>
- UniProt
<http://www.uniprot.org/uniprot/O75718>

Sources for This Summary

- Barnes AM, Chang W, Morello R, Cabral WA, Weis M, Eyre DR, Leikin S, Makareeva E, Kuznetsova N, Uveges TE, Ashok A, Flor AW, Mulvihill JJ, Wilson PL, Sundaram UT, Lee B, Marini JC. Deficiency of cartilage-associated protein in recessive lethal osteogenesis imperfecta. *N Engl J Med*. 2006 Dec 28;355(26):2757-64.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/17192541>
- OMIM: CARTILAGE-ASSOCIATED PROTEIN
<http://omim.org/entry/605497>
- Labuda M, Morissette J, Ward LM, Rauch F, Lalic L, Roughley PJ, Glorieux FH. Osteogenesis imperfecta type VII maps to the short arm of chromosome 3. *Bone*. 2002 Jul;31(1):19-25.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/12110407>
- Marini JC, Cabral WA, Barnes AM, Chang W. Components of the collagen prolyl 3-hydroxylation complex are crucial for normal bone development. *Cell Cycle*. 2007 Jul 15;6(14):1675-81. Epub 2007 May 18. Review.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/17630507>
- Morello R, Bertin TK, Chen Y, Hicks J, Tonachini L, Monticone M, Castagnola P, Rauch F, Glorieux FH, Vranka J, Bächinger HP, Pace JM, Schwarze U, Byers PH, Weis M, Fernandes RJ, Eyre DR, Yao Z, Boyce BF, Lee B. CRTAP is required for prolyl 3- hydroxylation and mutations cause recessive osteogenesis imperfecta. *Cell*. 2006 Oct 20;127(2):291-304.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/17055431>

- Tonachini L, Morello R, Monticone M, Skaug J, Scherer SW, Cancedda R, Castagnola P. cDNA cloning, characterization and chromosome mapping of the gene encoding human cartilage associated protein (CRTAP). *Cytogenet Cell Genet.* 1999;87(3-4):191-4.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/10702664>
 - Ward LM, Rauch F, Travers R, Chabot G, Azouz EM, Lalic L, Roughley PJ, Glorieux FH. Osteogenesis imperfecta type VII: an autosomal recessive form of brittle bone disease. *Bone.* 2002 Jul;31(1):12-8.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/12110406>
-

Reprinted from Genetics Home Reference:
<https://ghr.nlm.nih.gov/gene/CRTAP>

Reviewed: November 2007

Published: March 21, 2017

Lister Hill National Center for Biomedical Communications
U.S. National Library of Medicine
National Institutes of Health
Department of Health & Human Services